CDKN2B-AS1

Genome-wide association studies (GWAS) have revealed that common variants on or near EDNRA, HDAC9, SOX17, RP1, CDKN2B-AS1, and RBBP8 genes are associated with intracranial aneurysm (IA) in European or Japanese populations. However, due to population heterogeneity, whether these loci are associated with IA pathogenesis in Chinese individuals is still unknown. The purpose of this study was to investigate associations among GWAS-identified loci and risk of IA in a Chinese population.

MATERIALS AND METHODS: A total of 765 individuals (including 230 IA patients and 535 controls) were involved in this study. Twelve single nucleotide polymorphisms (SNPs) of candidate loci were genotyped using the Sequenom MassARRAY platform. Associations were analyzed using univariate or multivariate logistic regression analysis.

RESULTS: SNPs in CDKN2B-AS1 (especially rs10757272) showed significant associations with IA in dominant and additive models [odds ratio (OR), 2.99 and 1.43; 95% confidence interval (CI), 1.44-6.24 and 1.10-1.86, respectively]. A SNP near HDAC9 (rs10230207) was associated with IA in the dominant model (OR, 1.42; 95% CI, 1.01-1.99). One SNP near RP1 (rs1072737) showed a protective effect on IA in the dominant model (OR, 0.66; 95% CI, 0.46-0.95), while another SNP in RP1 (rs9298506) showed a risk effect on IA in a recessive model (OR, 3.82; 95% CI, 1.84-7.91). No associations were observed among common variants near EDNRA, SOX17, or RBBP8 and IA.

CONCLUSION: These data partially confirmed earlier results and showed that variants in CDKN2B-AS1, RP1, and HDAC9 could be genetic susceptibility factors for IA in a Chinese population [1)].

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Li B, Hu C, Liu J, Liao X, Xun J, Xiao M, Yan J. Associations among Genetic Variants and Intracranial Aneurysm in a Chinese Population. Yonsei Med J. 2019 Jul;60(7):651-658. doi: 10.3349/ymj.2019.60.7.651. PubMed PMID: 31250579; PubMed Central PMCID: PMC6597466.